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United States Patent [19]

Skolnick et al.

[11] **Patent Number:** 5,747,282[45] **Date of Patent:** May 5, 1998[54] **17Q-LINKED BREAST AND OVARIAN
CANCER SUSCEPTIBILITY GENE**

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[21] **Appl. No.:** 483,554[22] **Filed:** Jun. 7, 1995**Related U.S. Application Data**

[63] Continuation-in-part of Ser. No. 409,305, Mar. 24, 1995, abandoned, which is a continuation-in-part of Ser. No. 348,824, Nov. 29, 1994, abandoned, which is a continuation-in-part of Ser. No. 308,104, Sep. 16, 1994, which is a continuation-in-part of Ser. No. 300,266, Sep. 2, 1994, abandoned, which is a continuation-in-part of Ser. No. 289,221, Aug. 12, 1994, abandoned.

[51] **Int. Cl.⁶** C12P 21/06; C12Q 1/68; C07H 21/04

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[58] **Field of Search** 536/23.1, 23.5, 536/24.33; 435/69.1, 6, 7.1, 91.1, 91.2, 375, 320.1; 514/44

[56] **References Cited
PUBLICATIONS**

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[57] **ABSTRACT**

The present invention relates generally to the field of human genetics. Specifically, the present invention relates to methods and materials used to isolate and detect a human breast and ovarian cancer predisposing gene (BRCA1), some mutant alleles of which cause susceptibility to cancer, in particular breast and ovarian cancer. More specifically, the invention relates to germline mutations in the BRCA1 gene and their use in the diagnosis of predisposition to breast and ovarian cancer. The present invention further relates to somatic mutations in the BRCA1 gene in human breast and ovarian cancer and their use in the diagnosis and prognosis of human breast and ovarian cancer. Additionally, the invention relates to somatic mutations in the BRCA1 gene in other human cancers and their use in the diagnosis and prognosis of human cancers. The invention also relates to the therapy of human cancers which have a mutation in the BRCA1 gene, including gene therapy, protein replacement therapy and protein mimetics. The invention further relates to the screening of drugs for cancer therapy. Finally, the invention relates to the screening of the BRCA1 gene for mutations, which are useful for diagnosing the predisposition to breast and ovarian cancer.

20 Claims, 18 Drawing Sheets